American Journal of Medical Genetics 63:290–292 (1996)

Oculo-Facio-Cardio-Dental (OFCD) Syndrome

Robert J. Gorlin, Amir H. Marashi, and Hugo L. Obwegeser

University of Minnesota, Minneapolis (R.J.G.) and Private Practice, Oral and Maxillofacial Surgery, Mankato, Minnesota (A.H.M.); and Schwerzenbach, Switzerland (H.L.O.)

Oculo-facio-cardio-dental syndrome consists of (1) eye anomalies: congenital cataract and microphthalmia, or secondary glaucoma, (2) facial abnormalities: (long narrow face, high nasal bridge, pointed nose with cartilages separated at the tip, cleft palate, or submucous cleft palate, (3) cardiac anomalies: atrial septal defect (ASD), ventricular septal defect (VSD), or floppy mitral valve; and (4) dental abnormalities: canine radiculomegaly, delayed dentition, oligodontia, persistent primary teeth, or variable root length. Other less common findings are: sensorineural hearing loss, septate vagina, and syndactyly of toes 2-3. Inheritance may be an X-linked dominant trait, lethal in the male.

KEY WORDS: oculo-facio-cardio-dental syndrome, congenital cataracts, microphthalmia, ASD, VSD, canine radiculomegaly, X-linked dominant inheritance

INTRODUCTION

Marashi and Gorlin [1990, 1992] described three examples of congenital cataracts with radiculomegaly. They cited an earlier example of Hayward [1980] in which canine radiculomegaly, delayed dentition, persistent primary teeth, oligodontia, and congenital cataracts were found in an 18-year-old women. They also noted a 20-year-old female with marked canine radiculomegaly on whom further information was not available.

Wilkie et al. [1993] reported on a mother and daughter with congenital cataract, microphthalmia, and septal heart defect, suggesting that this combination represented a new syndrome. The mother's face was long and narrow, the philtrum was long, and the nose had a high bridge. In side view, the nose appeared pointed. The tip was broad with well-delineated cartilages. Eye findings included congenital cataract, secondary glaucoma, microphthalmia, and horizontal nystagmus. In the daughter, posterior embryotoxin was noted, and there was oligodontia with persistent primary teeth. The columella was broad. In the mother, the teeth had variable root length and canine radiculomegaly. The mother had ASD, and the daughter atrial septal defect and ventricular septal defect (ASD/VSD). Cutaneous syndactyly of toes 2–3 was observed in the daughter.

Aalfs et al. [1996] described 2 unrelated female patients with long narrow face, microphthalmia, microcornea, congenital cataracts, high nasal bridge, short nose with broad tip, long philtrum, sensorineural hearing loss, persistent primary teeth, oligodontia, ASD, VSD, and cutaneous syndactyly of toes 2–3. One of the 2 patients had cleft palate, and the other had septate vagina.

CLINICAL REPORTS

Case 1

We elected to reexamine a female patient first seen at age 20 years [Marashi and Gorlin, 1990] for routine dental examination.

The patient, now 26 years old, was born at term from an unremarkable pregnancy. Her brother and parents are otherwise unremarkable. The parents are first cousins. Birth weight was 3,500 g, and birth length 50 cm. Occipitofrontal circumference (OFC) was not recorded but was unremarkable.

Fig. 1. Patient with congenital cataracts and secondary glaucoma.

Received for publication November 6, 1995; no revisions.

Address reprint requests to Robert J. Gorlin, D.D.S., University of Minnesota School of Dentistry, 515 Delaware St. S.E., Room 16-127, Minneapolis, MN 55455.

Dedicated to Jürgen W. Spranger on the occasion of his 65th birthday with admiration and best wishes.

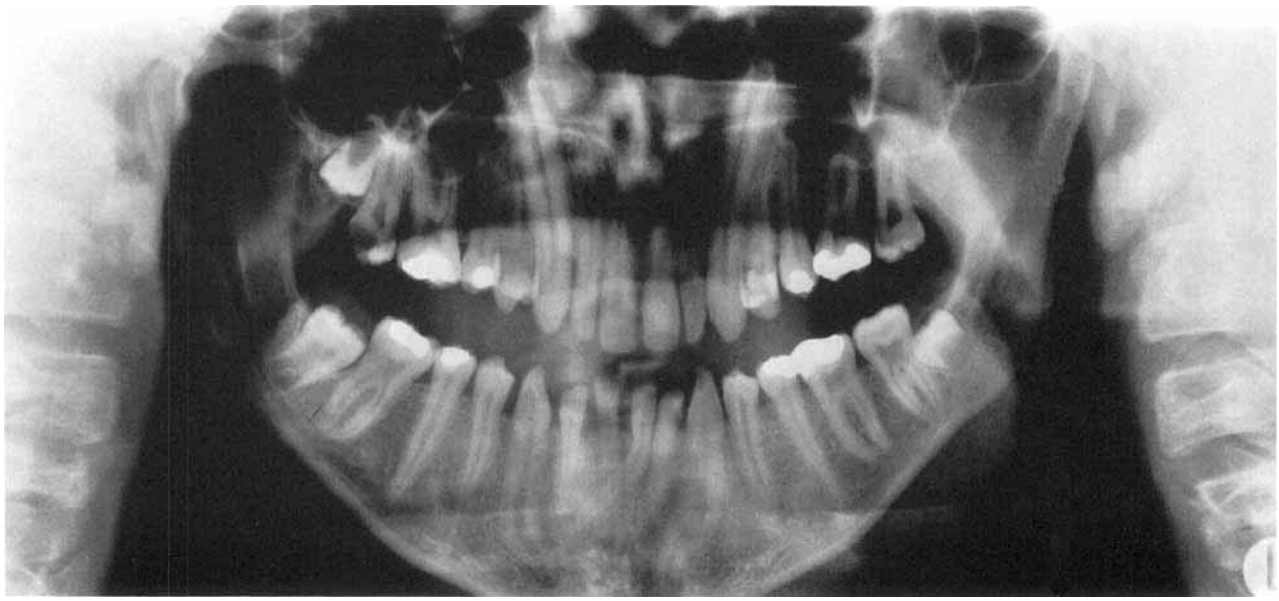

Fig. 2. X-ray of patient. Note remarkable length of teeth, especially roots of canine teeth which are still open (canine radiculomegaly).

At birth, cataracts, microcornea, and microphthalmia were evident. Secondary glaucoma developed which has resulted in clinical blindness (20/200). The patient has a narrow face, flat midface, and nose with high nasal bridge broad tip with cartilagenous clefting (Fig. 1). Results of a hearing test were unremarkable. A panoramic radiograph showed multiple root anomalies in all four quadrants, root dilaceration, and abnormally long-rooted canine teeth with open apices (Fig. 2). The incisors also had open apices. Mandibular asymmetry, extreme overbite, malocclusion and mild retrognathia were evident.

A grade III systolic ejection murmur was heard which on echocardiographic study, proved to be mitral valve prolapse. A soft-tissue syndactyly on toes 2–3 was evident bilaterally. Oligodontia and persistent primary teeth with dilaceration and canine radiculomegaly were noted in all four quadrants. Bifid uvula, submucous cleft palate, and mild bifid tongue tip were noted. Intelligence is normal.

Case 2

A female patient, now 36 years old, was referred at age 18 years for extraction of the canine teeth which exhibited extreme radicular enlargement. In addition, she was noted to have congenital cataracts bilaterally and submucous cleft palate.

Recent reexamination revealed moderate mental retardation. She suffers from recurrent bouts of vertigo. The cartilages of the nasal tip are separated. Syndactyly of toes 2-3 was evident. Congenital heart disease was not found and gynecologic examination of the patient was refused by her caretaker.

Radiographs of the jaws greatly resembled those seen in Figure 2. Several teeth were extracted. There was generalized radiculomegaly (Fig. 3). The root apices of the upper and lower canines reached the orbit and lower mandibular border, respectively. The root apices of the canine teeth had not closed at 18 years (Fig. 4).

DISCUSSION

The occurrence of this syndrome in two generations and in 7 females but not in males suggests that this syndrome is an X-linked dominant trait, lethal in the male.

The data derived from the case reports were not collected uniformly. In a few cases cited by Marashi and Gorlin [1990, 1992], the material consisted only of dental biopsy reports.

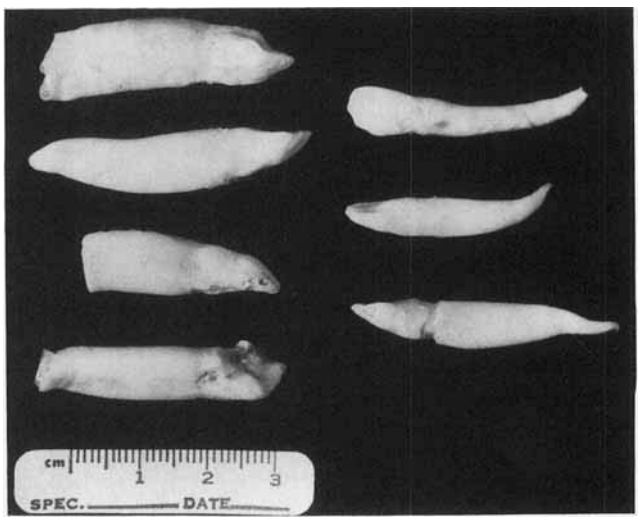

Fig. 3. Generalized radiculomegaly.

Fig. 4. Canine radiculomegaly with open apex.

Patients with this syndrome appear to have a long narrow face which is characterized by a sharp nose with a clearly defined bifid tip. A high nasal bridge was noted in at least 5 patients.

Eye changes evident at birth consist of congenital cataract and microphthalmia, or microcornea with resultant or secondary glaucoma.

The most unmistakable finding is canine radiculomegaly. This is not manifest orally, but its unique nature is seen on a panoramic radiograph of the jaws. The root ends of the canine teeth do not close until adulthood, the roots continuing to grow until the orbit and lower border of the mandible are reached by the maxillary and mandibular canines, respectively. (In some cases) this dental anomaly is often accompanied by oligodontia and persistence of primary teeth.

Cardiac anomalies of various types (ASD, VSD, and mitral valve prolapse) have been documented in at least 5 patients.

Cleft palate or submucous cleft palate was documented in 3 of the 7 patients.

Cutaneous soft-tissue syndactyly of toes 2–3 was found in 4 of the 7.

Miscellaneous findings have included septate vagina in one patient, and sensorineural hearing loss in another.

We would like to suggest the name oculo-facio-cardio-dental (OFCD) syndrome for the condition.

REFERENCES

Aalfs CM, Oosterwijk JC, von Schooneveld MJ, Begman CJ, Wabeke KB, Hennekam RCM (1996): A combination of ocular, facial, dental, cardiac, and audiological features in two unrelated adult females with normal intelligence: Confirmation of a syndrome. Clin Dysmorphol (in press).

Hayward JR (1980): Cuspid gigantism. Oral Surg Oral Med Oral Pathol 49:500-501.

Marashi AH, Gorlin RJ (1990): Cataracts and radiculomegaly of canines. Oral Surg Oral Med Oral Pathol 70:802-803.

Marashi AH, Gorlin RJ (1992): Radiculomegaly of canine teeth and congenital cataracts: Confirmation of a syndrome. Am J Med Genet 42:143.

Wilkie AOM, Taylor D, Seambler PJ, Baraitser M (1993): Congenital cataract, microphthalmia and septal heart defect in two generations: A new syndrome? Clin Dysmorphol 2:114-119.